GPSA
GENERAL PRACTICE SUPERVISION AUSTRALIA

Genomics in general practice

Genetic testing for human disease has never been more accessible, nor sought after by health consumers. Genetics counselling and testing is no longer solely the domain of specialist units, but increasingly becoming a routine element of Australian general practice. GPs can order a variety of different genetic tests, but direct-to-consumer marketing means that GPs are being asked to interpret complex tests that they may not have requested. There are a number of common genetic disorders (e.g. hereditary haemochromatosis) that registrars need to competently assess and manage. Furthermore, registrars increasingly need to consider a genetic contribution to a wide range of patient presentations, as well as develop a best practice approach to genetic counselling and test interpretation.

TEACHING AND LEARNING AREAS		• Common genetic disorders in general practice • How to take and interpret a comprehensive family history using a validated tool like the Family History Screening Questionnaire • Common scenarios involving genetics counselling and testing e.g. carrier and prenatal screening, developmental problems, cancer etc. • Indications and local pathways for genetics referral
PRE- SESSION ACTIVITIES		• Read the 2014 AFP article Genetics in General Practice • Review the resources at the NSW Centre for Genetics Education
TEACHING TIPS AND TRAPS		• Genomics is not the same as genetics! • Routinely taking a comprehensive family history is a fundamental element of good clinical practice and essential to identify patients at risk • There are important clinical, financial and ethical considerations when ordering genetic tests, including implications for notifying relatives and insurance companies • Consider Fragile X genetic testing and a chromosomal microarray in children with intellectual delay or developmental delay • Use appropriate risk assessment tools when discussing genetic testing for bowel, breast, ovarian and prostate cancers • Don't undertake genetic testing for MTHFR or APOE where the clinical utility is low – Choosing Wisely recommendation • Do not undertake genetic testing as a screening test for coeliac disease – Choosing Wisely recommendation
RESOURCES	**Read**	• RACGP Red Book 9th edition – Chapter 2 – Genetic counselling and testing • Australian Prescriber article 2017 – Retail Genetics • AFP article 2014 – Genetic Testing – Medicolegal Issues • RACGP resource – Genomics in General Practice • RACGP Advances in Genomic Testing
	Listen	• Australian Prescriber Podcast on Retail Genetics • RACP Episode 20 Genomics for the Generalist - Part 1 • RACP Episode 21 Genomics for the Generalist - Part 2
	Watch	• TED talk 2011 – Welcome to the genomic revolution
FOLLOW UP/ EXTENSION ACTIVITIES		• Registrar to develop a teaching session on genomics in general practice, including discussion on the enhanced use of routine, comprehensive family history taking in the practice • Registrar to undertake the role play with supervisor

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Clinical Reasoning Challenge

INSTRUCTIONS FOR SUPERVISOR

You are Hannah, a 42-year-old project manager, who has come to the doctor because your sister, aged 44, has just been diagnosed with breast cancer. You have been attending the practice for about 20 years.

"My sister has just been diagnosed with breast cancer and I wanted to see whether I should have any tests done"

Story

- You have just found out that your sister, aged 44, has breast cancer. She found a lump and it has been confirmed as a cancer. You don't really know much more except that she needs surgery.
- You feel completely well and have not felt any lumps.
- You have never had any breast problems.
- You have never had a mammogram.
- On questioning, you state that your mother died of ovarian cancer aged 64. You are not aware of any other breast or ovarian cancers in the family.
- You are Australian of European ancestry.
- You are quite worried and upset for your sister. You wonder whether you should have a mammogram and gene tests.
- You also have a 12 year old daughter and wonder what the future implications are for her.

Information in clinical record

- Occupation – Project manager
- Non-smoker
- Alcohol – 2 glasses wine/night
- No allergies
- PMH – nil significant
- No regular medications
- Social – married, 2 children
- FHx – not recorded

Assess

- Communication skills –
- History taking, especially FHx –
- Appropriate information seeking – use of risk assessment tool –
- Management plan –
- Follow-up –

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INSTRUCTIONS TO CANDIDATE

Hannah is a 42-year-old who has been attending the practice for about 20 years.

You have eight (8) minutes to:
- Take a focused history
- Outline management plan

Health summary
- Occupation – Project manager
- Non-smoker
- Alcohol – 2 glasses wine/night
- No allergies
- PMH – nil significant
- No regular medications
- Social – married, 2 children
- FHx – not recorded

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